GOVERNMENT OF INDIA
HEALTH AND FAMILY WELFARE
LOK SABHA

UNSTARRED QUESTION NO:2285
ANSWERED ON:05.12.2014
RARE ORPHAN DISEASES AND DISORDERS
Rai Shri Prem Das

Will the Minister of HEALTH AND FAMILY WELFARE be pleased to state:

(a) whether the Government has undertaken any study/ survey to assess the prevalence of various types of rare/ orphan diseases and disorders across the country, if so, the details and the outcome thereof, State/UT-wise;

(b) whether the Government has taken any steps to build awareness about such disease across the country and if so, the details thereof;

(c) the details of steps taken/proposed to be taken by the Government for improving the development and delivery of affordable diagnostics and treatments for rare disease in the country;

(d) whether the Government proposes to develop registries of conclusively diagnosed patients of rare/orphan diseases and disorders to improve treatment support; and

(e) if so, the details thereof along with the other steps being taken by the Government in this regard?

Answer

THE MINISTER OF HEALTH AND FAMILY WELFARE (SHRI JAGAT PRAKASH NADDA)

(a) to (c): While there is no nation-wide study conducted to assess the prevalence of various types of rare/orphan diseases and disorders across the country, Indian Council of Medical Research (ICMR) has conducted a National Task Force (NTF) Study on Newborn Screening for Congenital Hypothyroidism (CH) and Congenital Adrenal Hyperplasia (CAH) involving screening newborns in 5 regional centres at Delhi, Mumbai, Hyderabad, Chennai and Kolkata covering 1 lakh newborns. In this study, it was found that overall the prevalence of CH was 1 in 1130 and CAH was 1 in 5762. Treatment has been initiated on all affected children of the 5 regional centres.

ICMR has also conducted / supported workshops and prepared Clinical & Laboratory manuals as follows:-

(i) Hands on workshops on clinical, laboratory and data entry aspects. (ii) Workshop on Quality Control Methods. (iii) International Workshop: The Scenes and Art of Newborn Screening. (iv) Launch of an interactive website www.icmrmetbionetindia.org (v) Preparation of advocacy material in 8 languages. (vi) Printing of Laboratory Manual. (vii) Printing of ICMR priced publication "Clinical Manual for Inborn Errors of Metabolism".

(d) & (e) : There is no proposal at present to develop registries. However, a study entitled "Creating a web based directory of genetic services and genetic information in Indian (www.geneticsindia.com) is being supported by ICMR for a platform for rare genetic disorders.